What is Hypertrophic Cardiomyopathy (HCM)?

HCM is an inherited heart disease and the leading cause of sudden cardiac death in young adults. HCM is the most common inherited cardiovascular disease, affecting about 1 individual in 500. Hypertrophic cardiomyopathy means “overgrowth of the heart muscle.” In most cases, the thickening of the heart muscle is limited to one part of the heart (the left ventricle). While big muscles are usually associated with strength, the bigger HCM heart is weaker, not stronger, than a normal heart and often cannot pump blood effectively. In addition, the thickening of the heart may lead to disturbances in the heart’s rhythm. In about 10% of patients, HCM can lead to premature death from severe complications such as heart failure, stroke, or cardiac arrest. HCM is the most common heart-related cause of sudden death in young adults and responsible for at least one third of all sudden death seen among competitive athletes.

What causes Hypertrophic Cardiomyopathy?

Most cases of HCM are caused by a gene defect. Most cases of HCM are due to a genetic defect, which can occur in any one of several different genes. About half of all individuals who harbor an HCM-associated defect in their genes will develop HCM. All individuals who harbor an HCM-associated genetic defect will pass on this defect to about half of their children.

What are signs of Hypertrophic Cardiomyopathy?

The first signs of HCM may be fainting, exercise intolerance, or fatigue. HCM symptoms most commonly first occur in adolescence, but can appear at any age. The first signs of HCM often include fainting, exercise intolerance, or fatigue. Young athletes with HCM most often complain of chest pain and shortness of breath during exercise. In some HCM patients, a doctor may hear a heart murmur.

How is Hypertrophic Cardiomyopathy diagnosed?

HCM is usually diagnosed by an echocardiogram. HCM is usually diagnosed by an echocardiogram – a test that allows the doctor to see the thickening of the heart. In athletes, the diagnosis by echocardiogram may be more difficult since rigorous athletic training can lead to “athlete’s heart,” a mild thickening of the heart that is not related to HCM. Most patients with HCM also have changes in their heart’s rhythm that can be seen with a test called an electrocardiogram (EKG).

Is everybody with Hypertrophic Cardiomyopathy at risk of Sudden Cardiac Death?

Only patients with certain risk factors are at high risk of sudden cardiac death from HCM. Only about 10 to 20% of patients with HCM are at a high risk for sudden death. High-risk patients can typically be identified by the presence of certain risk factors, such as an abnormally fast resting heart rate (called ventricular tachycardia), severe thickening of the heart, abnormal blood pressure response during exercise, or a history of fainting. All patients with HCM should be regularly checked for these risk factors.
How can genetic testing help families with Hypertrophic Cardiomyopathy?

Genetic testing can help to identify family members who are at increased risk of HCM and family members who are not.

Sudden death due to HCM most commonly occurs in adolescents, who often are not aware of their condition. It is therefore important to find out if an adolescent (or child or young adult) is genetically predisposed to HCM, so that regular heart exams can be performed. Family history of HCM is a strong indicator of risk, even before any symptoms develop.

Genetic testing can identify the genetic defect that leads to HCM in a particular family. Other members of the same family can then easily be tested for presence of this genetic defect (called the “familial mutation”). Family members who harbor the familial mutation should undergo regular heart exams, consider preventative lifestyle changes, and seek treatment early. Family members who do not harbor the familial mutation no longer have to worry about developing the disease or passing it on to their children. Neither they nor their children need to undergo the rigorous and costly heart exams recommended for HCM patients.

How is Hypertrophic Cardiomyopathy treated?

HCM is treated with medication, surgery, or implantation of a cardioverter-defibrillator.

Treatment for HCM depends on the patient’s symptoms and the risk of sudden death. Congestive heart failure, or loss in the heart’s pumping power, is treated with drugs called beta-blockers and, in some patients, by surgical removal of extra tissue from the thickened part of the heart.

Some disturbances in the heart’s rhythm may be treated with medication or by implantation of a pacemaker. Other rhythm disturbances may require implantation of an electrical device called an implantable cardioverter-defibrillator (ICD), which has been shown to prevent sudden death in high-risk patients.

More information is available from:

Correlagen Diagnostics
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Hypertrophic Cardiomyopathy Association (HCMA)
www.4hcm.org
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Children’s Cardiomyopathy Foundation (CCF)
www.childrenscardiomyopathy.org
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C.A.R.E. Foundation
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References